

Extra Chromosome, Extra Love

Fazladan Kromozom, Fazladan Sevgi

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ABSTRACT

Objective: Down syndrome is the most common chromosomal disorder and characterized by dysmorphic features, intellectual disability and frequent developmental abnormalities. Though the syndrome is a lifetime health issue for society, the awareness of our community on the syndrome is not sufficient.

Methods: This study is planned to define the level of awareness of our population on simple Down syndrome facts. A questionnaire consisting sociodemographic characteristics of the participants and Down syndrome facts was applied to 283 randomly selected participants.

Results: The majority of the participants (89.8%) have heard of Down syndrome and 70% of the participants were aware that Down syndrome can be diagnosed during pregnancy. Even though less than half of them (42.4%) was clued in frequent association of congenital malformations with the syndrome, more participants (55.1%) knew that the advanced maternal age was a risk factor. There were statistically significant differences between different sociodemographic groups on awareness of particular subjects.

Conclusion: Even though the certain demographics were well informed about the syndrome, there are still excessive amount of prejudice against Trisomy 21 patients; and therefore, more social projects are needed to raise the awareness.

Key Words: Down Syndrome; Trisomy 21; Health Knowledge; Attitudes

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ÖZET

Amaç: Toplumda en yaygın gözlenen kromozomal hastalık olan Down Sendromu dismorfik bulgular, entelektüel yetersizlik ve sık olarak da gelişimsel anomalilerle seyrederek. Sendrom yaşam boyu süren sağlık problemlerinden olsa da, hakkındaki farkındalık seviyesi toplumumuzda yetersizdir.

Yöntem: Çalışma toplumumuzun basit Down sendromu gerçekleri hakkındaki farkındalık seviyesini belirlemek amacıyla planlandı. 283 randomize seçilmiş katılımcıya sosyodemografik özelliklerini ve Down sendromu bilgilerini içeren bir anket uygulandı.

Bulgular: Katılımcıların çoğunluğu (%89,8) Down sendromunu duymuştu ve %70'i sendromun gebelik sürecinde tespit edilebileceğini biliyordu. Katılımcıların yarısından azı (%42,4) konjenital malformasyonlarla sendromun sık ilişkisinden haberdarken, daha fazla sayıda katılımcı (%55,1) ileri anne yaşının bir risk faktörü olduğunu biliyordu. Belirli konularda farklı sosyodemografik grupların farkındalığı arasında istatistiksel olarak belirgin bir fark vardı.

Sonuç: Toplumun bazı sosyodemografik grupları sendrom hakkında yüksek farkındalığa sahip olsa da, Trizomi 21 vakalarına karşı hala oldukça fazla bir önyargı olduğu gözlenmekte; ve bu da sendrom hakkındaki farkındalığı yükseltmek için daha fazla sosyal projelere ihtiyaç olduğu göstermektedir.

Anahtar Sözcükler: Down sendromu; Trizomi 21; Sağlık Bilgisi; Tutum

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INTRODUCTION

Down Syndrome (Trisomy 21) is the most common autosomal chromosomal disorder in humans. It is caused by the total and/or partial trisomy of the chromosome 21. Approximately 1 in 792 children in United States was born with Down Syndrome (1). Even though the clinical features of the disease were described more than a century ago (2), the genetic cause of the disease was found in 1959 (3). Since then, clinical features and genetic baseline of the syndrome have been defined and being elaborated by scientists.

Dysmorphic features, intellectual disability and congenital heart disease are among the most frequent features of Trisomy 21. The patients can be recognized shortly after birth by dysmorphic features including upslanting palpebral fissures, epicanthal folds, protruding tongue and single transverse palmar crease. The average intelligence quotient (IQ) of people with Down syndrome is between 20-70, and the mean IQ is approximately 50 (4,5). Nearly half of them are diagnosed with congenital heart disease and it is the leading cause of mortality (6). In addition to these; short stature, congenital cataract, congenital gastrointestinal system anomalies, seizures, hearing impairment, thyroid disorders and early onset demantia are frequent features of the syndrome (7).

Down syndrome has different types of cytogenetic forms. The standart (regular type) Trisomy 21 is the most common subtype (~95%), and it is usually caused by maternal meiotic errors. Advanced maternal age is presumed to predispose meiotic errors and it is one of the most remarkable major risk factors for Down syndrome. Mosaic Down syndrome, which results from postzygotic mitotic errors, accounts for approximately 2% of the individuals and present with milder phenotypes. Isochromosome Trisomy 21 is the second most common form, and the syndrome may also be caused by translocations or other rare rearrangements. There are different risks for recurrence on each group for families with a history of Down syndrome (8).

Because that the form of chromosomal abnormality effects the phenotypical spectrum and the recurrence risks, detection of genetic anomaly at the earliest stage is very important for families seeking for genetic consultation. High risk pregnancies for Down syndrome can be determined by prenatal screening tests and the definite diagnosis can be made by prenatal diagnosis tests. Therefore families can make a decision on the pregnancy and predicting the potential phenotypical features and the level of intellectual disability are necessary to anticipate the need for medical support, follow-up, and educational needs of the individuals to develop special social skills and integrate into society.

Studies have shown that there is still an unawareness on basic facts of Down syndrome and prenatal screening/diagnosis opportunities in our population. It is extremely important to create public awareness to eliminating this lack of information. Therefore, in this study, we conducted a survey to measure the approach to Down syndrome of our patients.

METHODS

The study was carried out on the Down Syndrome Awareness stand established at the entrance of the Eskisehir Osmangazi University (ESOGU) Hospital on March 21, World Down Syndrome Day.

A questionnaire consisting of 16 questions was applied to the volunteers who referred to the ESOGU Hospital for various reasons (see questionnaire form). The participants were randomly selected and all of them were over the age of 18.

Oral consent was obtained with the approval of the ethical committee numbered 57-2018. An informative brochure about Trisomy 21 was distributed to the volunteers once they completed the questionnaire.

The questionnaire consisted of two parts; 1: a series of questions evaluating sociodemographic characteristics of the participants (Table 1) 2: questions about Down syndrome (Table 2). The sociodemographic questions included the age, gender, education and employment status of the participants. The questions about Down syndrome were about some simple Down syndrome facts, and the translation of questionnaire is presented at supplementary materials.

The data obtained as a result of the questionnaire were evaluated statistically with chi square test in IBM SPSS statistic 15 program.

RESULTS

There were 283 participants in the study. The age distribution ranged from 18 to 81 years (mean 40.37 ± 15.05 years). While the most participants (43.1%) were between 30-50 years old, 75 of them (26.5%) were 51 or older. 163 individuals (57.6%) were female and 120 (42.7%) were male. 60.4% of the participants stated that they were employed. Most participants were undergraduate or completed an university education (41.3%) but 36.7% of them only attended primary school. The collective sociodemographic characteristics of the individuals participating the study are given in the Table 1.

Table 1: Sociodemographic characteristics of the participants

Demographics	n	%	
Age (years)	18-30	86	30.4
	31-50	122	43.1
	51 and over	75	26.5
Gender	Female	163	57.6
	Male	120	42.7
Employment	Unemployed	112	39.6
	Employed	171	60.4
Education Status	Primary education	104	36.7
	High school	62	21.9
	Undergraduate +	117	41.3
n : Number		% : Percentage	

The first question about the syndrome was whether the participants have ever heard of the syndrome (see questionnaire form). Even though the majority of the participants (89.8%) said that they have heard of Down syndrome, and more than a quarter (28.3%) stated that they know someone with the syndrome, rate of participants who have never heard about it (10.2%) was more than our expectations. It was also found that 70% of the participants knew that Down syndrome can be diagnosed during pregnancy by prenatal testing (Table 2). There were statistically significant differences between different sociodemographic groups on this subject: Women were more aware than men, people who applied to university were more aware than the ones attended primary or secondary schools ($p < 0.001$ ve $p < 0.001$), and also participants aged 51 and over were statistically significantly less conscious than younger ($p < 0.0001$).

Table 2: The answers of question 6: “Do you know that diagnosis of Down Syndrome can be made by prenatal tests during pregnancy?” according to sociodemographic characteristics of the participants.

Demographics / Answers		Yes	(n / %)	No	(n / %)	Total	(n / %)	X ² ; p
Age (years)								
	18-30	68 (79.1)		18(20.9)		86 (%30.4)		
	31-50	92 (75.4)		30 (24.6)		122 (43.1)		18.406; <0.001
	51 and over	38 (50.7)		37 (49.3)		75 (26.5)		
Gender								
	Female	130 (79.8)		33 (20.2)		163 (57.6)		17.532; <0.001
	Male	68 (56.7)		52 (43.3)		120 (42.4)		
Education Status								
	Primary education	55 (52.9)		49 (47.1)		104 (36.7)		38.439; <0.001
	High school	38 (61.3)		24 (38.7)		62 (21.9)		
	Undergraduate +	105 (89.7)		12 (10.3)		117 (41.3)		
Employment								
	Employed	116 (67.8)		55 (32.2)		171 (60.4)		0.931; >0.05
	Unemployed	82 (73.2)		30 (26.8)		112 (39.6)		
n: number	%: percentage	X ² : chi-square		p: p-value				

While a great majority of the participants (88%) stated that they can recognize people with Down syndrome when they see them, less than half of them (42.4%) was clued in frequent association of congenital malformations with the syndrome. 70% of the participants knew that individuals with Down syndrome can integrate into society by special educations. But unfortunately, 14.1% said that they were prejudiced against Down syndrome and 7.4% of them even believed that Down syndrome might be contagious.

More than half of the participants (55.1%) knew that the advanced maternal age was a risk factor (Table 3) and approximately forty four percentage of the participants declared that the recurrence risk could be increased because of a previous affected child (Table 4). There was no significant difference in different gender, age and employment groups for awareness of the association between advanced maternal age or previous pregnancies with trisomy 21 and Down syndrome risk. But participants with higher educations were more conscious on this concern (p = 0.001).

Table 3: Answers of question 9: “Do you know that advanced maternal age confers higher risk for Down syndrome?” according to sociodemographic characteristics of the participants.

Demographic / Answers		Yes (%)	(n / %)	Unaware (n / %)	Total (n / %)	X ² ; p
Age (years)						
	18-30	56 (65.1)		30 (34.9)	86 (30.4)	
	31-50	66 (54.1)		56 (45.9)	122 (43.1)	6.429; >0.05
	51 and over	34 (45.3)		41 (54.7)	75 (26.5)	>0.05
Gender						
	Female	96 (58.9)		67 (41.1)	163 (57.6)	2.211; >0.05
	Male	60 (50)		60 (50)	120 (42.4)	
Education						
	Primary education	42 (40.4)		62 (59.6)	104 (36.7)	
	High school	25 (40.3)		37 (59.7)	62 (21.9)	35.372; <0.001
	Undergraduate+	89 (76.1)		28 (23.9)	117 (41.3)	<0.001
Employment						
	Employed	94 (55)		77 (45)	171 (60.4)	0.004; >0.05
	Unemployed	62 (55.4)		50 (44.6)	112 (39.6)	>0.05
n: number	%: percentage	X ² : chi-square		p: p-value		

Table 4: Answers of question 15: “Do you know that a previous pregnancy history of Down syndrome increases the recurrence risk?” according to sociodemographic characteristics of the participants.

Demographics / Answers		Yes (n / %)	No (n / %)	Total (n / %)	X ² ; p
Age					
	18-30	46 (53.5)	40 (46.5)	86 (30.4)	
	31-50	50 (41)	72 (59)	122 (43.1)	4.946; >0.05
	51 and over	28 (37.3)	47 (62.7)	75 (26.5)	
Gender					
	Female	72 (44.2)	91 (55.8)	163 (57.6)	0.020; >0.05
	Male	52 (43.3)	68 (56.7)	120 (42.4)	
Education					
	Primary education	37 (35.6)	67 (64.4)	104 (36.7)	
	High school	20 (32.3)	42 (67.7)	62 (21.9)	14.829; <0.001
	Undergraduate+	67 (57.3)	50 (42.7)	117 (41.3)	<0.001
Employment					
	Employed	78 (45.6)	93 (54.4)	171 (60.4)	
	Unemployed	46 (41.1)	66 (58.9)	112 (39.6)	0.567; >0.05
n: Number	%: Percentage	X ² : chi-square		p: p-value	

DISCUSSION

Down syndrome is a genetic disorder with well described clinical features. Prenatal screening tests can estimate the approximate risk for Down syndrome during pregnancies and invasive prenatal tests can establish the diagnosis. Prenatal screening and diagnosis tests not only aim to confer healthy pregnancies to populations but also provide a range of informed choice to families at risk of having a child with an abnormality. A multicenter study in our country evaluated 6041 prenatal samples and it was reported that just as expected, Trisomy 21 was the most common anomaly with a frequency of 48% (9). The high rate of Trisomy 21 on pregnancies proves that it is necessary to inform our community on this topic.

The present study was planned to investigate the current awareness on Down syndrome. The most striking finding was that there was a significant difference between different age, gender, education groups on awareness of prenatal diagnosis of Down syndrome. The tests were known more by women compared to men, and highly educated group compared to others ($p<0.001$ and $p<0.001$) and less known by people aged 51 years or older ($p<0.0001$) (Table 2). It would be reasonable to consider that young people knows more about prenatal diagnosis of Down syndrome and risk of advanced maternal age because it is easier for them to reach more information regarding these matters thanks to technological developments.

It was inevitable that age of motherhood has been increasing with women getting better education. Although advanced maternal age is one of the major reasons that families are seeking for prenatal diagnosis (9), 44.9% of our participants were unaware of its risk. It was also seen that participants who attended university had more awareness on prenatal diagnosis, advanced maternal age being a risk factor, and families with previous pregnancies of Trisomy 21 had higher risk for recurrence. There is also another study, in which awareness on, application for and attitudes towards prenatal screening tests of Turkish women were investigated (10). The study revealed that women who have lower education levels, lower incomes, who are unemployed, raised in patriarchal families, who had consanguineous marriages and who does not have personal clinicians were less aware and used prenatal screening tests less commonly (10). The conclusions of the researchers supported our outcomes.

Another study advocates that funding clinics taking care of Down syndrome patients helps to elevate health status of children and adolescents with the syndrome (11). It is also shown that, in addition to special educations, interacting children with normal neuromotor development on routine education programs encourages children with Trisomy 21 to evolve their social skills (12). Previous studies proves that it is necessary for Down syndrome patients to get special educations and be accepted by society in order to adapt them. Seventy percent of the participants of this study was conscious of the fact that Down syndrome patients can integrate into society by special educations. It is known that prejudices against Down syndrome reduces life quality of the patients (13). Due to the rate of 14% of the participants prejudiced against the syndrome and 7.4% participants thought that it was contagious, our awareness activity in our society with the slogan of "extra chromosome, extra love" revealed the need of more social projects about Down syndrome.

A limitation of our study is that, the participants are the patients who applied to our hospital. It would be more convenient to plan a population based study. The higher awareness of Down syndrome in younger ages and higher education group interpreted that, by the effect of social media and reaching easy information sources, younger and highly educated participants were more informed on this issue. Raising awareness on all of the demographics will make

it easier to find better solutions on medical and social issues, increase life quality of patients with Down syndrome and their families, and it will be easier for them to integrate into society. Therefore, we suggest that more educative sessions or social projects are needed for all social groups to raise awareness on this subject.

Conflict of interest

No conflict of interest was declared by the authors.

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World Down Syndrome Day, Down Syndrome Awareness Questionnaire

1. Age:
2. Sex:
 - a. Female
 - b. Male
3. Education:
 - a. Primary school
 - b. Middle school
 - c. High school
 - d. Undergraduate
 - e. Postgraduate
4. Employment:
 - a. Employed
 - b. Unemployed
5. Have you ever heard Down syndrome?
 - a. Yes
 - b. No
6. Do you know that diagnosis of Down Syndrome can be made by prenatal tests during pregnancy?
 - a. Yes
 - b. No
7. Do you know that there is no definite cure for Down syndrome, but patients can integrate into society by special educations?
 - a. Yes
 - b. No
8. Do you think that Down syndrome is contagious?
 - a. Yes
 - b. No
9. Do you know that advanced maternal age confers higher risk for Down syndrome?
 - a. Yes
 - b. No
10. Can you recognize Down syndrome patients if you see them?
 - a. Yes
 - b. No
11. Do you know that today, March 21st, is celebrated as World Down Syndrome Day?
 - a. Yes
 - b. No
12. Do you have any prejudices (like they are dangerous or like they are aggressive) against individuals with Down syndrome, even just a little?
 - a. Yes
 - b. No
13. Do you realize that people with Down syndrome smiles so beautifully?
 - a. Yes
 - b. No
14. Do you know that in addition to intellectual disability, cardiac anomalies or other system abnormalities can be seen on Down syndrome?
 - a. Yes
 - b. No
15. Do you know that a previous pregnancy history of Down syndrome increases the recurrence risk?
 - a. Yes
 - b. No
16. Do you personally know anyone with Down syndrome? If so, how is your communication?
 - a. Yes.....
 - b. No